22q11 DELETION SYNDROME IN IRELAND – REVIEW OF A NEWLY ESTABLISHED DEDICATED CLINIC

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Background

22q11 Deletion Syndrome is the most common microdeletion syndrome in humans. The prevalence is estimated to be 1 in 2000-4000¹. Affected individuals may present with a diverse range of medical conditions. Care for this population is frequently fragmented.

A need for a dedicated clinic in Ireland was identified by both medical professionals caring for these children and their families through 22q Ireland parent support group. Currently the following specialists have arranged dedicated clinic slots for children with 22q11DS on Tuesdays to improve care co-ordination: Immunology, Child Psychiatry, Plastic Surgery and General Developmental Paediatrics. The Consensus Document on 22q11DS (Max Appeal) guidelines were used as a reference².

Objective

Our aim is to establish if a dedicated clinic can identify and address unmet needs within this population using a newly designed intake form to record information.

Methods

The intake forms and letters of all new referrals seen in the first six months were reviewed. Previous specialty input along with referrals made and investigations requested were recorded. Parents reported unmet needs and wishes were also recorded.

Results

During the initial 6 months of the 22q11DS Clinic 17 children were assessed. The attendance rate was 94%. The age ranged from 8 months to 14 years.

Children attended from 8 counties around Ireland. 18% of children had an affected relative(s).

Results continued

Parents reported a range of unmet needs and there was huge variation in previous access to services. Parents main reported desires (in 35%) was co-ordination and planning of care, along with same-day appointments.

100% of the school-age children were in mainstream schools, 57% of those had access to a special needs assistant. 12% were currently in preschool and were getting additional support.

The following investigations were requested: Blood testing in 71%, renal ultrasound in 29%.

Referrals were made in 53% of cases, across 13 different specialties.

29% of children had their appointment co-ordinated with another specialist on the same day.

Conclusions

In this population co-ordinated care and a dedicated clinic has been shown in other centres to be associated with reduced morbidity and improved quality of life³. It is hoped that we can improve this clinic further with a dedicated nurse specialist and improve care co-ordination further by engaging other specialists to run clinics on the same day.

Literature cited

¹ McDonald-McGinn and Sullivan. Chromosome 22q11.2 Deletion Syndrome. Medicine. 90(1):1-18, JAN 2011

²Max Appeal! Consensus Document Development Committee. Consensus Document on 22q11 Deletion Syndrome (22q11DS). 2017

³ Alex Habel, Beverly Tsai-Goodman et al. Towards a safety net for management of 22q11.2 deletion syndrome: guidelines for our times. Eur J Pediatr. 2014; 173(6): 757–765.